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Tay-Sachs Disease

What Is It?

Tay-Sachs disease is an inherited disease caused by an abnormal gene. People with tl abnormal gene cannot properly break down a fatty material called ganglioside GM2. T material builds up in the brain, eventually damaging nerve cells and causing neurolog problems.

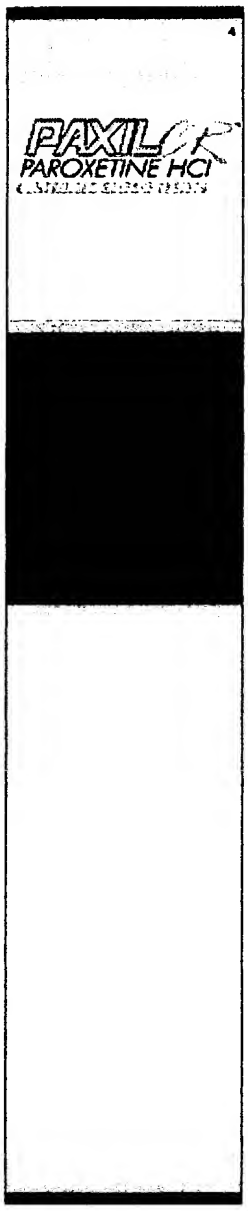
Infants usually begin to show signs of the disease between 3 and 6 months of age. Cl with Tay-Sachs disease can become deaf, blind, paralyzed, and usually die by the age

Tay-Sachs disease is an autosomal recessive inherited disorder, where a child inherits of this abnormal gene from each parent. The parent does not actually have the diseas carries the Tay-Sachs gene and passes it on to the baby. If both parents have the abı Tay-Sachs gene, there is a one-in-four chance that the child will inherit the gene from them and have Tay-Sachs disease. Tay-Sachs disease is more common in Ashkenazi J about 1 in 30 people with this ancestry carry a copy of the gene.

Three types of related conditions are often included in the definition of Tay-Sachs dise because they affect the same gene.

- A juvenile form usually appears between ages 2 and 5. The symptoms resemble classic Tay-Sachs disease, and death usually occurs by age 15.
- A chronic form usually appears by age 5. Symptoms resemble the classic form, milder, with resulting muscle weakness, slurred speech, tremors and (sometime illness.
- An adult form (called late-onset Tay-Sachs, or LOTS) resembles the chronic forı appears much later in life, between the teens and the 30s.

Other names for Tay-Sachs disease include Tay-Sachs sphingolipidosis; infantile gang



lipidosis; cerebromacular degeneration; GM2 gangliosidosis, Type 1; and amaurotic fa infantile idiocy.

Symptoms

Early signs and symptoms of Tay-Sachs disease may include:

- Loss of muscle tone
- Exaggerated response to sudden noises
- Lack of energy
- Loss of motor skills, such as the ability to roll over, crawl, reach for things or sit

In its advanced form, the disease causes a gradual loss of vision, deafness, seizures, paralysis and dementia. Red spots may appear on the retina.

Diagnosis

Tay-Sachs disease can be diagnosed before birth, through amniocentesis or chorionic sampling. If you are considering having a child, you and your partner can have a bloo see if you are carrying the Tay-Sachs disease gene. After birth, your doctor can do a to see if your child has Tay-Sachs disease and to rule out similar neurological conditic

Expected Duration

Tay-Sachs disease is a lifelong condition.

Prevention

Genetic counseling before conception can help both parents understand what the chai that they will have a child with Tay-Sachs disease. If both parents-to-be are carriers a mother is pregnant, amniocentesis or chorionic villus sampling can determine if the fe affected.

Treatment

There is no effective treatment for Tay-Sachs disease. As with other fatal diseases, tr is aimed at relieving symptoms and making the child and family comfortable.

When To Call A Professional

Any child or adult showing neurological impairment should be evaluated by a doctor.

Prognosis

Classic infantile Tay-Sachs disease is a fatal disease. The long-term prognosis for the form is not known.

Additional Info

National Institute f Neur l gical Disorders and Stroke
P.O. Box 5801
Bethesda, MD 20824
Phone: (301) 496-5751

http://www.ninds.nih.gov/

National Organization for Rare Disorders, Inc.
P.O. Box 8923
New Fairfield, CT 06812-8923
Phone: (203) 746-6518
Toll-Free: (800) 999-6673
Fax: (203) 746-6481
http://www.rarediseases.org/

March of Dimes Birth Defects Foundation
1275 Mamaroneck Ave.
White Plains, NY 10605
Toll-Free: (888) 663-4637
http://www.modimes.org/

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